



## DOCK6 gene

dedicator of cytokinesis 6

### Normal Function

The *DOCK6* gene provides instructions for making a protein known as a guanine nucleotide exchange factor (GEF). GEFs turn on (activate) proteins called GTPases, which play an important role in chemical signaling within cells. Often referred to as molecular switches, GTPases can be turned on and off. GTPases are turned off (inactivated) when they are attached (bound) to a molecule called GDP and are activated when they are bound to another molecule called GTP. The DOCK6 protein activates GTPases known as Cdc42 and Rac1 by exchanging GTP for the attached GDP. Once Cdc42 and Rac1 are active, they transmit signals that are critical for various aspects of embryonic development. The DOCK6 protein appears to regulate these GTPases specifically during development of the limbs, skull, and heart. DOCK6 also plays a role in the development of fibers (axons) that extend from nerve cells.

### Health Conditions Related to Genetic Changes

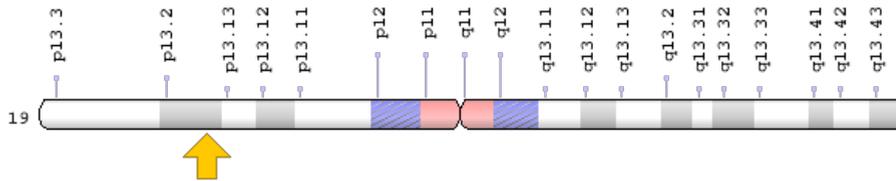
#### Adams-Oliver syndrome

Mutations in the *DOCK6* gene cause Adams-Oliver syndrome, a condition characterized by areas of missing skin (aplasia cutis congenita), usually on the scalp, and malformations of the hands and feet. Neurological abnormalities, such as brain or eye malformations and intellectual disability, are more common in DOCK6-related Adams-Oliver syndrome than in cases associated with other genes. Most *DOCK6* gene mutations involved in this condition lead to production of an abnormally short DOCK6 protein that is likely unable to function. Other mutations change single protein building blocks (amino acids) in the DOCK6 protein, which impairs the protein's normal function. The inability of DOCK6 to turn on Cdc42 or Rac1 leads to a reduction in their signaling, which impairs proper development of certain tissues, including the skin on the top of the head and the bones in the hands and feet.

## Chromosomal Location

Cytogenetic Location: 19p13.2, which is the short (p) arm of chromosome 19 at position 13.2

Molecular Location: base pairs 11,199,293 to 11,262,502 on chromosome 19 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- AOS2
- dedicator of cytokinesis protein 6
- DOCK6\_HUMAN
- KIAA1395
- ZIR1

## Additional Information & Resources

### Educational Resources

- Madame Curie Bioscience Database (2000): Rho GTPases Function as Membrane-Associated GDP/GTP-Regulated Molecular Switches  
[https://www.ncbi.nlm.nih.gov/books/NBK6594/#\\_A39189\\_](https://www.ncbi.nlm.nih.gov/books/NBK6594/#_A39189_)

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28DOCK6%5BTIAB%5D%29+OR+%28%28AOS2%5BTIAB%5D%29+OR+%28ZIR1%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5BIa%5D+AND+human%5Bmh%5D>

### OMIM

- DEDICATOR OF CYTOKINESIS 6  
<http://omim.org/entry/614194>

## Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_DOCK6.html](http://atlasgeneticsoncology.org/Genes/GC_DOCK6.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=DOCK6%5Bgene%5D>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=19189](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=19189)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/57572>
- UniProt  
<http://www.uniprot.org/uniprot/Q96HP0>

## **Sources for This Summary**

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*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/25824905>

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